CBS gene

cystathionine-beta-synthase

Normal Function

The *CBS* gene provides instructions for making an enzyme called cystathionine beta-synthase. This enzyme acts in a chemical pathway and is responsible for using vitamin B6 to convert building block of proteins (amino acid) called homocysteine and serine to a molecule called cytathionine. Another enzyme then converts cystathionine to the amino acid cysteine, which is used to build proteins or is broken down and excreted in urine. Additionally, other amino acids, including methionine, are produced in this pathway.

Health Conditions Related to Genetic Changes

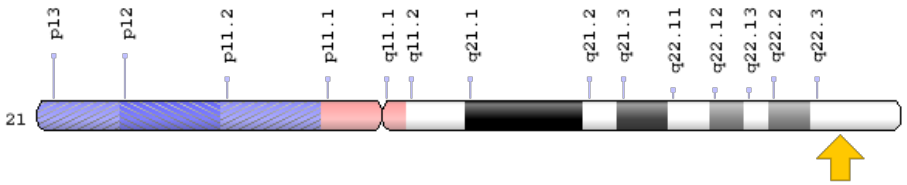
homocystinuria

More than 150 mutations that cause homocystinuria have been identified in the *CBS* gene. Most of these mutations change single amino acids in cystathionine beta-synthase. The most common mutation substitutes the amino acid threonine for the amino acid isoleucine at position 278 in the enzyme (written as Ile278Thr or I278T). Another common mutation, which is the most frequent cause of homocystinuria in the Irish population, replaces the amino acid glycine with the amino acid serine at position 307 (written as Gly307Ser or G307S). These mutations disrupt the normal function of cystathionine beta-synthase. As a result, homocysteine and other potentially toxic compounds build up in the blood, and homocysteine is excreted in urine. Researchers have not determined how excess homocysteine leads to the signs and symptoms of homocystinuria.

Chromosomal Location

Cytogenetic Location: 21q22.3, which is the long (q) arm of chromosome 21 at position 22.3

Molecular Location: base pairs 43,053,190 to 43,076,868 on chromosome 21 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- beta-thionase
- CBS_HUMAN
- HIP4
- methylcysteine synthase
- serine sulfhydrase

Additional Information & Resources

GeneReviews

- Homocystinuria Caused by Cystathionine Beta-Synthase Deficiency https://www.ncbi.nlm.nih.gov/books/NBK1524

Genetic Testing Registry

- GTR: Genetic tests for CBS https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=875%5Bgeneid%5D

Scientific articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28CBS%5BTIAB%5D%29+OR+%28cystathionine-beta-synthase%5BTIAB%5D%29%29+AND+%28%28l-serine+hydro-lyase+%28adding+homocysteine%29%29+OR+%28beta-thionase%5BMAJR%5D%29+OR+%28serine+sulfhydrase%5BMAJR%5D%29+OR+%28cystathionine+synthetase%5BMAJR%5D%29+OR+%28cystathionine+beta-synthase%5BMAJR%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D

OMIM

- CYSTATHIONINE BETA-SYNTHASE
  http://omim.org/entry/613381

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_CBS.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=CBS%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=1550
- Kraus Lab at the University of Colorado Health Sciences Center
  http://www.ucdenver.edu/academics/colleges/medicalschool/programs/kraus/Pages/home.aspx
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/875
- UniProt: CBS_HUMAN
  http://www.uniprot.org/uniprot/P35520
- UniProt: CBSL_HUMAN
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